



SMARCAD1 gene

SWI/SNF-related, matrix-associated actin-dependent regulator of chromatin, subfamily a, containing DEAD/H box 1

Normal Function

The *SMARCAD1* gene provides instructions for making two versions (isoforms) of the *SMARCAD1* protein: a full-length isoform and a shorter, skin-specific isoform. The full-length isoform is active (expressed) in multiple tissues, where it regulates the activity of a wide variety of genes involved in maintaining the stability of cells' genetic information. The skin-specific isoform is expressed only in skin cells, and little is known about its function. However, it appears to play a critical role in the formation of dermatoglyphs, which are the patterns of skin ridges on the pads of the fingers that form the basis for each person's unique fingerprints. These ridges are also present on the toes, the palms of the hands, and the soles of the feet. Dermatoglyphs develop before birth and remain the same throughout life. The activity of the skin-specific isoform of the *SMARCAD1* protein is likely one of several factors that determine each person's unique fingerprint pattern.

Health Conditions Related to Genetic Changes

adermatoglyphia

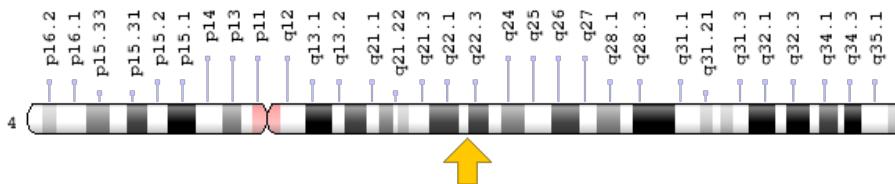
At least four mutations in the *SMARCAD1* gene have been found to cause adermatoglyphia, which is the absence of dermatoglyphs on the hands and feet. Because affected individuals do not have skin ridges on the pads of their fingers, they cannot be identified on the basis of their fingerprints. Adermatoglyphia can occur without any related signs and symptoms, or it may be associated with other features, typically affecting the skin.

The mutations that cause adermatoglyphia affect the skin-specific isoform of the *SMARCAD1* protein but not the full-length isoform. These genetic changes prevent the production of any functional skin-specific isoform from one copy of the gene, which reduces the total amount of this protein in skin cells. Although it is unclear how these genetic changes cause adermatoglyphia, researchers speculate that a shortage of the skin-specific version of the *SMARCAD1* protein impairs signaling pathways needed for normal skin development and function, including the formation of dermatoglyphs.

Chromosomal Location

Cytogenetic Location: 4q22.3, which is the long (q) arm of chromosome 4 at position 22.3

Molecular Location: base pairs 94,207,608 to 94,291,292 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ADERM
- ATP-dependent helicase 1
- DKFZP762K2015
- ETL1
- HEL1
- KIAA1122
- SMRCD_HUMAN

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SMARCAD1%5BTIAB%5D%29+OR+%28HEL1%5BTI%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- SWI/SNF-RELATED, MATRIX-ASSOCIATED ACTIN-DEPENDENT REGULATOR OF CHROMATIN, SUBFAMILY A, DEAD/H BOX-CONTAINING, 1
<http://omim.org/entry/612761>

Research Resources

- **Atlas of Genetics and Cytogenetics in Oncology and Haematology**
http://atlasgeneticsoncology.org/Genes/GC_SMARCAD1.html
- **ClinVar**
<https://www.ncbi.nlm.nih.gov/clinvar?term=SMARCAD1%5Bgene%5D>
- **HGNC Gene Symbol Report**
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18398
- **NCBI Gene**
<https://www.ncbi.nlm.nih.gov/gene/56916>
- **UniProt**
<http://www.uniprot.org/uniprot/Q9H4L7>

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